IFWO

RAW SEQUENCE LISTING             DATE: 09/17/2004
PATENT APPLICATION:  US/10/766,760        TIME: 15:44:58

Input Set : **A:\Sequence Listing.TXT**
Output Set: **N:\CRF4\09172004\J766760.raw**

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   4 <110> APPLICANT: Klein, Michael
   5        Schultz, Dennis
   7 <120> TITLE OF INVENTION: Gene Mutation Associated with
   8        Age-Related Macular Degeneration
  11 <130> FILE REFERENCE: 49321-117
  13 <140> CURRENT APPLICATION NUMBER: 10/766,760
C--> 14 <141> CURRENT FILING DATE: 2004-01-27
  16 <150> PRIOR APPLICATION NUMBER: 60/443,214
  17 <151> PRIOR FILING DATE: 2003-01-27
  19 <160> NUMBER OF SEQ ID NOS: 186
  21 <170> SOFTWARE: FastSEQ for Windows Version 4.0
  23 <210> SEQ ID NO: 1
  24 <211> LENGTH: 18206
  25 <212> TYPE: DNA
  26 <213> ORGANISM: Homo sapiens
  28 <220> FEATURE:
  29 <221> NAME/KEY: CDS
  30 <222> LOCATION: (230)...(17137)
  32 <221> NAME/KEY: variation
  33 <222> LOCATION: (14197)...(14197)
  34 <223> OTHER INFORMATION: A and G allelic variation exists at this position
W--> 36 <221> variation
  37 <222> LOCATION: (17811)...(17811)
  38 <223> OTHER INFORMATION: C and T allelic variation exists at this position
W--> 40 <400> 1
  41 gaagccgcat ccagacaaaa gctgccgcat ccctgccctg cccaaccccct ggagggattc 60
  42 gagtttggtg cttgtccccg tctgattctc agcgccaaac tttttgctag ttcagagatt 120
  43 ccaagagtct gatgagttac tctgagagga aaccctctgc ctgttgttga ggaggactga 180
  44 gcacagtgct taggcgctga gggggaaaaa gaggggggaaa aaaaagaaa atg att tcc 238
  45                                                        Met Ile Ser
  46                                                         1
  48 tgg gaa gtt gtc cat aca gta ttc ctg ttt gct ctt ctt tat tct tcc    286
  49 Trp Glu Val Val His Thr Val Phe Leu Phe Ala Leu Leu Tyr Ser Ser
  50   5                10                15
  52 cta gct caa gat gcg agc ccc cag tca gag atc aga gct gag gaa att    334
  53 Leu Ala Gln Asp Ala Ser Pro Gln Ser Glu Ile Arg Ala Glu Glu Ile
  54  20                25                30                35
  56 ccc gag ggg gcc tcc acg ttg gct ttt gtg ttt gat gtg act ggt tct    382
  57 Pro Glu Gly Ala Ser Thr Leu Ala Phe Val Phe Asp Val Thr Gly Ser
  58                40                45                50
  60 atg tat gat gat tta gtt cag gtg att gaa ggg gct tcc aaa att ttg    430
  61 Met Tyr Asp Asp Leu Val Gln Val Ile Glu Gly Ala Ser Lys Ile Leu
  62                55                60                65
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 64 gag acg tct ttg aaa aga cct aaa aga cct ctt ttc aac ttt gcg ttg     478
 65 Glu Thr Ser Leu Lys Arg Pro Lys Arg Pro Leu Phe Asn Phe Ala Leu
 66             70              75              80
 68 gtg cct ttc cat gat cca gaa att ggc cca gtg aca att acc aca gat     526
 69 Val Pro Phe His Asp Pro Glu Ile Gly Pro Val Thr Ile Thr Thr Asp
 70         85              90              95
 72 ccc aag aaa ttt caa tat gaa ctc aga gaa ctg tat gtt cag ggt ggt     574
 73 Pro Lys Lys Phe Gln Tyr Glu Leu Arg Glu Leu Tyr Val Gln Gly Gly
 74 100             105             110             115
 76 ggt gat tgc cca gaa atg agt att gga gct ata aaa att gcc ttg gaa     622
 77 Gly Asp Cys Pro Glu Met Ser Ile Gly Ala Ile Lys Ile Ala Leu Glu
 78             120             125             130
 80 att tct ctt cct ggt tct ttc atc tat gtt ttc act gat gct cgg tcc     670
 81 Ile Ser Leu Pro Gly Ser Phe Ile Tyr Val Phe Thr Asp Ala Arg Ser
 82             135             140             145
 84 aaa gat tac cgg ctc acc cat gag gtg ctg caa ctt atc caa cag aaa     718
 85 Lys Asp Tyr Arg Leu Thr His Glu Val Leu Gln Leu Ile Gln Gln Lys
 86             150             155             160
 88 cag tca caa gtc gta ttt gtt ctg act gga gat tgt gat gac agg acc     766
 89 Gln Ser Gln Val Val Phe Val Leu Thr Gly Asp Cys Asp Asp Arg Thr
 90         165             170             175
 92 cat att gga tat aaa gtc tat gaa gaa att gcc tct aca agt tct ggt     814
 93 His Ile Gly Tyr Lys Val Tyr Glu Glu Ile Ala Ser Thr Ser Ser Gly
 94 180             185             190             195
 96 caa gtg ttc cat ctg gac aaa aaa caa gtt aat gag gta tta aaa tgg     862
 97 Gln Val Phe His Leu Asp Lys Lys Gln Val Asn Glu Val Leu Lys Trp
 98             200             205             210
100 gta gaa gaa gca gta cag gcc tcc aaa gtt cac ctt tta tcc aca gat     910
101 Val Glu Glu Ala Val Gln Ala Ser Lys Val His Leu Leu Ser Thr Asp
102             215             220             225
104 cat ttg gaa cag gct gta aat act tgg aga att cct ttt gat ccc agc     958
105 His Leu Glu Gln Ala Val Asn Thr Trp Arg Ile Pro Phe Asp Pro Ser
106             230             235             240
108 ctg aaa gag gtc act gtg tct ttg agt ggg cct tct cca atg att gaa    1006
109 Leu Lys Glu Val Thr Val Ser Leu Ser Gly Pro Ser Pro Met Ile Glu
110         245             250             255
112 att cgc aat cct tta ggg aag ctg ata aaa aag gga ttt ggc ctg cat    1054
113 Ile Arg Asn Pro Leu Gly Lys Leu Ile Lys Lys Gly Phe Gly Leu His
114 260             265             270             275
116 gag cta tta aat atc cat aac tct gcc aaa gta gtg aat gtg aaa gag    1102
117 Glu Leu Leu Asn Ile His Asn Ser Ala Lys Val Val Asn Val Lys Glu
118             280             285             290
120 cca gag gct gga atg tgg aca gtg aag acc tca agc agt gga agg cac    1150
121 Pro Glu Ala Gly Met Trp Thr Val Lys Thr Ser Ser Ser Gly Arg His
122             295             300             305
124 tct gtt cgc att act ggc ctc agt act att gat ttc cga gct ggc ttt    1198
125 Ser Val Arg Ile Thr Gly Leu Ser Thr Ile Asp Phe Arg Ala Gly Phe
126             310             315             320
128 tct cga aag ccc acc ctg gac ttc aaa aaa aca gtc agc aga cca gtg    1246
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129 Ser Arg Lys Pro Thr Leu Asp Phe Lys Lys Thr Val Ser Arg Pro Val
130     325             330             335
132 caa gga ata cct acc tat gta ctg ctc aat act tct gga att tcc act    1294
133 Gln Gly Ile Pro Thr Tyr Val Leu Leu Asn Thr Ser Gly Ile Ser Thr
134 340             345             350             355
136 cca gct aga ata gat ctt ctt gaa ctt ttg agt atc tca gga agt tct    1342
137 Pro Ala Arg Ile Asp Leu Leu Glu Leu Leu Ser Ile Ser Gly Ser Ser
138             360             365             370
140 ctt aag act att cct gtt aaa tat tac cca cat cga aaa cct tat ggc    1390
141 Leu Lys Thr Ile Pro Val Lys Tyr Tyr Pro His Arg Lys Pro Tyr Gly
142             375             380             385
144 ata tgg aat att tct gac ttt gta cca cca aat gaa gct ttc ttt ctc    1438
145 Ile Trp Asn Ile Ser Asp Phe Val Pro Pro Asn Glu Ala Phe Phe Leu
146             390             395             400
148 aaa gta aca ggc tat gat aaa gat gat tac ctc ttc cag aga gta tca    1486
149 Lys Val Thr Gly Tyr Asp Lys Asp Asp Tyr Leu Phe Gln Arg Val Ser
150     405             410             415
152 agt gtt tcc ttt tct agt att gtc cca gat gct ccc aaa gtt acg atg    1534
153 Ser Val Ser Phe Ser Ser Ile Val Pro Asp Ala Pro Lys Val Thr Met
154 420             425             430             435
156 cct gag aaa acc cca gga tac tat ctg cag ccg ggc caa att ccc tgc    1582
157 Pro Glu Lys Thr Pro Gly Tyr Tyr Leu Gln Pro Gly Gln Ile Pro Cys
158             440             445             450
160 tct gtt gac agt ctt ttg ccc ttt acc ttg agc ttt gtc aga aat gga    1630
161 Ser Val Asp Ser Leu Leu Pro Phe Thr Leu Ser Phe Val Arg Asn Gly
162             455             460             465
164 gtt aca ctt gga gta gac cag tat ttg aaa gaa tct gcc agt gtg aac    1678
165 Val Thr Leu Gly Val Asp Gln Tyr Leu Lys Glu Ser Ala Ser Val Asn
166             470             475             480
168 tta gat att gca aag gtc act ttg tct gac gaa ggt ttc tat gaa tgc    1726
169 Leu Asp Ile Ala Lys Val Thr Leu Ser Asp Glu Gly Phe Tyr Glu Cys
170     485             490             495
172 att gct gtc agc agt gca ggt act gga cgg gca cag aca ttt ttt gac    1774
173 Ile Ala Val Ser Ser Ala Gly Thr Gly Arg Ala Gln Thr Phe Phe Asp
174 500             505             510             515
176 gta tca gag ccc cct ccg gtc atc caa gtg cct aac aat gtt aca gtc    1822
177 Val Ser Glu Pro Pro Pro Val Ile Gln Val Pro Asn Asn Val Thr Val
178             520             525             530
180 act cct gga gag aga gca gtt tta aca tgt ctc atc atc agt gcg gtg    1870
181 Thr Pro Gly Glu Arg Ala Val Leu Thr Cys Leu Ile Ile Ser Ala Val
182             535             540             545
184 gat tac aat cta acc tgg cag agg aat gac aga gat gtc aga ctg gca    1918
185 Asp Tyr Asn Leu Thr Trp Gln Arg Asn Asp Arg Asp Val Arg Leu Ala
186             550             555             560
188 gag cca gcg aga att agg acc ttg gct aat ctg tca ttg gag cta aag    1966
189 Glu Pro Ala Arg Ile Arg Thr Leu Ala Asn Leu Ser Leu Glu Leu Lys
190     565             570             575
192 agt gtg aaa ttc aac gat gct gga gag tat cat tgt atg gtt tct agt    2014
193 Ser Val Lys Phe Asn Asp Ala Gly Glu Tyr His Cys Met Val Ser Ser
```

```
194 580                  585                  590                  595
196 gaa ggt gga tca tca gcc gct tca gtt ttc ctc aca gtg caa gaa cca    2062
197 Glu Gly Gly Ser Ser Ala Ala Ser Val Phe Leu Thr Val Gln Glu Pro
198                  600                  605                  610
200 ccc aaa gtc act gtg atg ccc aag aat cag tct ttc aca gga ggg tct    2110
201 Pro Lys Val Thr Val Met Pro Lys Asn Gln Ser Phe Thr Gly Gly Ser
202                  615                  620                  625
204 gag gtc tcc atc atg tgt tct gca aca ggt tat ccc aaa cca aag att    2158
205 Glu Val Ser Ile Met Cys Ser Ala Thr Gly Tyr Pro Lys Pro Lys Ile
206                  630                  635                  640
208 gcc tgg acc gtt aac gat atg ttt atc gtg ggt tca cac agg tat agg    2206
209 Ala Trp Thr Val Asn Asp Met Phe Ile Val Gly Ser His Arg Tyr Arg
210      645                  650                  655
212 atg acc tca gat ggt acc tta ttt atc aaa aat gca gct ccc aaa gat    2254
213 Met Thr Ser Asp Gly Thr Leu Phe Ile Lys Asn Ala Ala Pro Lys Asp
214 660                  665                  670                  675
216 gca ggg atc tat ggt tgc cta gca agt aat tca gct gga aca gat aaa    2302
217 Ala Gly Ile Tyr Gly Cys Leu Ala Ser Asn Ser Ala Gly Thr Asp Lys
218                  680                  685                  690
220 cag aat tct act ctc aga tac att gaa gcc cct aag ttg atg gta gtt    2350
221 Gln Asn Ser Thr Leu Arg Tyr Ile Glu Ala Pro Lys Leu Met Val Val
222                  695                  700                  705
224 cag agt gag ctc ttg gtt gcc ctt ggg gat ata acc gtt atg gaa tgc    2398
225 Gln Ser Glu Leu Leu Val Ala Leu Gly Asp Ile Thr Val Met Glu Cys
226           710                  715                  720
228 aaa acc tct ggt att cct cca cct caa gtt aaa tgg ttc aaa gga gat    2446
229 Lys Thr Ser Gly Ile Pro Pro Pro Gln Val Lys Trp Phe Lys Gly Asp
230      725                  730                  735
232 ctt gag ttg agg ccc tca aca ttc ctc att att gac cct ctc ttg gga    2494
233 Leu Glu Leu Arg Pro Ser Thr Phe Leu Ile Ile Asp Pro Leu Leu Gly
234 740                  745                  750                  755
236 ctt ttg aag att caa gaa aca caa gat ctg gat gct ggc gat tat acc    2542
237 Leu Leu Lys Ile Gln Glu Thr Gln Asp Leu Asp Ala Gly Asp Tyr Thr
238                  760                  765                  770
240 tgt gta gcc atc aat gag gct gga aga gca act ggc aag ata act ctg    2590
241 Cys Val Ala Ile Asn Glu Ala Gly Arg Ala Thr Gly Lys Ile Thr Leu
242                  775                  780                  785
244 gat gtt ggc tca cct cca gtt ttc ata caa gaa cct gct gat gtg tct    2638
245 Asp Val Gly Ser Pro Pro Val Phe Ile Gln Glu Pro Ala Asp Val Ser
246           790                  795                  800
248 atg gaa att ggc tca aat gtg aca tta cct tgt tat gtt cag ggt tat    2686
249 Met Glu Ile Gly Ser Asn Val Thr Leu Pro Cys Tyr Val Gln Gly Tyr
250      805                  810                  815
252 cca gaa cca aca atc aaa tgg cga aga tta gac aac atg cca att ttc    2734
253 Pro Glu Pro Thr Ile Lys Trp Arg Arg Leu Asp Asn Met Pro Ile Phe
254 820             825                  830                  835
256 tca aga cct ttt tca gtt agt tcc atc agc caa cta aga aca gga gct    2782
257 Ser Arg Pro Phe Ser Val Ser Ser Ile Ser Gln Leu Arg Thr Gly Ala
258                  840                  845                  850
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RAW SEQUENCE LISTING         DATE: 09/17/2004
PATENT APPLICATION:  US/10/766,760     TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT
Output Set: N:\CRF4\09172004\J766760.raw

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260 ctc ttt att tta aac tta tgg gca agt gat aaa gga acc tat att tgt   2830
261 Leu Phe Ile Leu Asn Leu Trp Ala Ser Asp Lys Gly Thr Tyr Ile Cys
262             855                 860                 865
264 gaa gct gaa aac cag ttt gga aag atc cag tca gag aca aca gta aca   2878
265 Glu Ala Glu Asn Gln Phe Gly Lys Ile Gln Ser Glu Thr Thr Val Thr
266         870                 875                 880
268 gtg acc gga ctt gtt gct cca ctt att gga atc agc cct tca gtg gcc   2926
269 Val Thr Gly Leu Val Ala Pro Leu Ile Gly Ile Ser Pro Ser Val Ala
270         885                 890                 895
272 aat gtt att gaa gga cag cag ctt act ttg ccc tgt act ctg tta gct   2974
273 Asn Val Ile Glu Gly Gln Gln Leu Thr Leu Pro Cys Thr Leu Leu Ala
274 900                 905                 910                 915
276 gga aat ccc att cca gaa cgt cgg tgg att aag aat tca gct atg ttg   3022
277 Gly Asn Pro Ile Pro Glu Arg Arg Trp Ile Lys Asn Ser Ala Met Leu
278             920                 925                 930
280 ctc caa aat cct tac atc act gtg cgc agt gat ggg agc ctc cat att   3070
281 Leu Gln Asn Pro Tyr Ile Thr Val Arg Ser Asp Gly Ser Leu His Ile
282             935                 940                 945
284 gaa aga gtt cag ctt cag gat ggt ggt gaa tat act tgt gtg gcc agt   3118
285 Glu Arg Val Gln Leu Gln Asp Gly Gly Glu Tyr Thr Cys Val Ala Ser
286         950                 955                 960
288 aac gtt gct ggg acc aat aac aaa act acc tct gtg gtt gtg cat gtt   3166
289 Asn Val Ala Gly Thr Asn Asn Lys Thr Thr Ser Val Val Val His Val
290         965                 970                 975
292 ctg cca acc att cag cat ggg cag cag ata ctc agt aca att gaa ggc   3214
293 Leu Pro Thr Ile Gln His Gly Gln Gln Ile Leu Ser Thr Ile Glu Gly
294 980                 985                 990                 995
296 att cca gta act tta cca tgc aaa gca agt gga aat ccc aaa ccg tct   3262
297 Ile Pro Val Thr Leu Pro Cys Lys Ala Ser Gly Asn Pro Lys Pro Ser
298             1000                1005                1010
300 gtc atc tgg tcc aag aaa gga gag ctg att tca acc agc agt gct aag   3310
301 Val Ile Trp Ser Lys Lys Gly Glu Leu Ile Ser Thr Ser Ser Ala Lys
302             1015                1020                1025
304 ttt tca gca gga gct gat ggt agt ctg tat gtg gta tca cct gga gga   3358
305 Phe Ser Ala Gly Ala Asp Gly Ser Leu Tyr Val Val Ser Pro Gly Gly
306         1030                1035                1040
308 gag gag agt ggg gag tat gtc tgc act gcc acc aat aca gcc ggc tac   3406
309 Glu Glu Ser Gly Glu Tyr Val Cys Thr Ala Thr Asn Thr Ala Gly Tyr
310     1045                1050                1055
312 gcc aaa agg aaa gtg cag cta aca gtc tat gta agg ccc aga gtg ttt   3454
313 Ala Lys Arg Lys Val Gln Leu Thr Val Tyr Val Arg Pro Arg Val Phe
314 1060                1065                1070                1075
316 gga gat caa cga gga ctg tcc cag gat aag cct gtt gag atc tcc gtc   3502
317 Gly Asp Gln Arg Gly Leu Ser Gln Asp Lys Pro Val Glu Ile Ser Val
318             1080                1085                1090
320 ctt gca ggg gaa gag gta aca ctt cca tgt gaa gtg aag agc tta cct   3550
321 Leu Ala Gly Glu Glu Val Thr Leu Pro Cys Glu Val Lys Ser Leu Pro
322         1095                1100                1105
324 cca ccc ata att act tgg gcc aaa gaa acc cag ctc atc tca ccg ttc   3598
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**VERIFICATION SUMMARY**                    DATE: 09/17/2004
PATENT APPLICATION:  **US/10/766,760**       TIME: 15:44:59

Input Set : **A:\Sequence Listing.TXT**
Output Set: **N:\CRF4\09172004\J766760.raw**

L:14 M:271 C: Current Filing Date differs, Replaced Current Filing Date
L:36 M:258 W: Mandatory Feature missing, <220> Tag not found for SEQ ID#:1
L:40 M:258 W: Mandatory Feature missing, <220> Tag not found for SEQ ID#:1
L:1460 M:336 W: Invalid Amino Acid Number in Coding Region, SEQ ID:1